GDF6 gene

growth differentiation factor 6

Normal Function

The *GDF6* gene provides instructions for making a protein that is part of the transforming growth factor beta (TGFβ) superfamily, which is a group of proteins that help control the growth and development of tissues throughout the body. Within the TGFβ superfamily, the GDF6 protein belongs to the bone morphogenetic protein family, which is involved in regulating the growth and maturation (differentiation) of bone and cartilage. Cartilage is a tough but flexible tissue that makes up much of the skeleton during early development. The proteins in this family are regulators of cell growth and differentiation both before and after birth. The GDF6 protein is necessary for the formation of bones and joints in the limbs, skull, spine, chest, and ribs. The protein is involved in setting up boundaries between bones during skeletal development.

The GDF6 protein has also been found to be involved in the development of the eyes, specifically the specialized light-sensitive tissue that lines the back of the eye called the retina. The GDF6 protein likely plays a role in the survival of specialized cells within the retina that detect light and color (photoreceptor cells).

Health Conditions Related to Genetic Changes

coloboma

Klippel-Feil syndrome

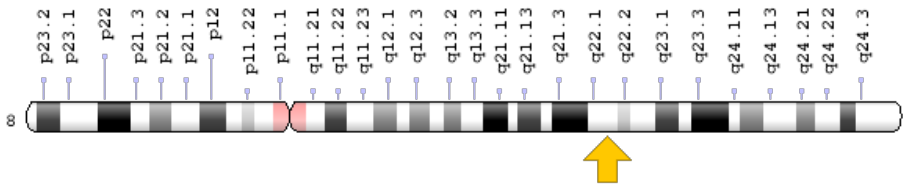
At least 10 mutations in the *GDF6* gene have been found to cause Klippel-Feil syndrome, a condition characterized by the abnormal joining (fusion) of two or more spinal bones in the neck (cervical vertebrae) and a variety of other features affecting many parts of the body. Most *GDF6* gene mutations that cause Klippel-Feil syndrome replace single protein building blocks (amino acids) in the GDF6 protein. These mutations likely lead to a reduction in functional protein. Although the GDF6 protein is involved in bone growth and the formation of vertebrae, it is unclear how a shortage of this protein leads to incomplete separation of the cervical vertebrae in people with Klippel-Feil syndrome.

microphthalmia

Chromosomal Location

Cytogenetic Location: 8q22.1, which is the long (q) arm of chromosome 8 at position 22.1

Molecular Location: base pairs 96,142,330 to 96,160,792 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BMP13
- CDMP2
- GDF-6
- GDF6_HUMAN
- growth/differentiation factor 6
- KFS
- KFS1
- SCDO4
- SGM1

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Bone Morphogenetic Proteins https://www.ncbi.nlm.nih.gov/books/NBK6520/#A46768

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GDF6%5BTIAB%5D%29+OR+%28growth+differentiation+factor+6%5BTIAB%5D%29%29+OR+%28BMP13%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

OMIM

- GROWTH/DIFFERENTIATION FACTOR 6
  http://omim.org/entry/601147

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GDF6.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GDF6%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4221
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/392255
- UniProt
  http://www.uniprot.org/uniprot/Q6KF10

Sources for This Summary

- Asai-Coakwell M, March L, Dai XH, Duval M, Lopez I, French CR, Famulski J, De Baere E, Francis PJ, Sundaresan P, Sauvé Y, Koenekoop RK, Berry FB, Allison WT, Waskiewicz AJ, Lehmann OJ. Contribution of growth differentiation factor 6-dependent cell survival to early-onset retinal dystrophies. Hum Mol Genet. 2013 Apr 1;22(7):1432-42. doi: 10.1093/hmg/dds560. Epub 2013 Jan 9.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23307924
- OMIM: GROWTH/DIFFERENTIATION FACTOR 6
  http://omim.org/entry/601147
- Portnoy ME, McDermott KJ, Antonellis A, Margulies EH, Prasad AB; NISC Comparative Sequencing Program, Kingsley DM, Green ED, Mortlock DP. Detection of potential GDF6 regulatory elements by multispecies sequence comparisons and identification of a skeletal joint enhancer. Genomics. 2005 Sep;86(3):295-305.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15979840

- Pregizer S, Mortlock DP. Control of BMP gene expression by long-range regulatory elements. Cytokine Growth Factor Rev. 2009 Oct-Dec;20(5-6):509-15. doi: 10.1016/j.cytogfr.2009.10.011. Epub 2009 Nov 8. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19900834
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2787762/
- Tassabehji M, Fang ZM, Hilton EN, McGaughran J, Zhao Z, de Bock CE, Howard E, Malass M, Donnai D, Diwan A, Manson FD, Murrell D, Clarke RA. Mutations in GDF6 are associated with vertebral segmentation defects in Klippel-Feil syndrome. Hum Mutat. 2008 Aug;29(8):1017-27. doi: 10.1002/humu.20741.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18425797

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/GDF6

Reviewed: May 2015
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services